



Figure 2

Mutation	Exemplary Pool	3' blocking group	Oligo type (Arm)	Sequence (5'-3')	ϵ_{900} M ⁻¹ cm ⁻¹
2789+5G>A	1	none	inverter	TTTGGTTGTGCTGTGGCTCTCTTGAAAGTGAT (SEQ ID NO:1)	330800
2789+5G>A	1	hex	probe/DM	CGCCGCGGAGATATCCATGTGCTATTGTG (SEQ ID NO:2)	306500
2789+5G>A	1	none	synthetic target	CATTACCAATAGCAATGGAAATTCATGCTCCAGGACGACACCAACCCAAA (SEQ ID NO:3)	687000
R1162X	1	none	inverter	GTTACCTGTGTGGCATGTCAATGAAGCTTAAAGACTCT (SEQ ID NO:4)	428000
R1162X	1	hex	probe/DM	CGCCGCGAGGAGCTCACAGTCG (SEQ ID NO:5)	253000
R1162X	1	none	synthetic target	TCAGATGCGATCTGTGAGCTGAGCTTTAAGTCTCATGAACTGCCAACAGAGGTTAAAC (SEQ ID NO:6)	659000
R347P	4	none	inverter	CAGGGAATTTGCCGAGTGACCGCCATGT (SEQ ID NO:7)	306800
R347P	4	hex	probe/ER24	ACGACGCGGAGGCGAGCAACAATGCGAG (SEQ ID NO:8)	318200
R347P	4	none	synthetic target	CTCATCTGCAATTTGTGCCCATGCGCGGTCTCGGCAATTCCTGTGGG (SEQ ID NO:9)	488000
1898+1G>A	1	hex	inverter	GACTCTCTTTTGATACCTTAGATGTTTAAACAGAAAATAATTTGAAAGT (SEQ ID NO:10)	619000
1898+1G>A	1	none	probe/DM	CGCCGCGAGGATATGTTCTTGTGAATACCTACTAT (SEQ ID NO:11)	386500
1898+1G>A	1	none	synthetic target	ATAAGAGAGATATCAAGAAACAATCTTTCAAAATATCTTTTCTGTGTTAAACATCTAGGTATCCAAAAGGAGAGTC (SEQ ID NO:12)	904800
2184delA	4	none	inverter	CGCCAAACTCTCCAGTCTGTAAAAAGATTTTTC (SEQ ID NO:13)	393000
2184delA	4	hex	probe/DM	CGCCGCGAGGGTTCTGCGAGGAGACA (SEQ ID NO:14)	305200
del1507	1	none	inverter	GCTTGATGACGCTGTGATCTATATTCATAGCAAAACACCAAT (SEQ ID NO:15)	509300
del1507	1	hex	probe/DM	CGCCGCGAGGAGATATTTCTTTAATGTGTGCC (SEQ ID NO:16)	345200
del1507	1	none	synthetic target	GCCTGGCAGGATATAAGAAATAATCTTGGTGTCTCTATGATGAATATAGATACAGAAAGCGTCATCAAAAGCATGCC (SEQ ID NO:17)	866800
G85E	4	none	inverter	GCCTCTGGCGGATGTTTTCTGGAGATTTATGTTCTGTG (SEQ ID NO:18)	409100
G85E	4	hex	probe/ER24	ACGACGCGGAGAAATCTTTTATATATAGGGTAA (SEQ ID NO:19)	431700
G85E	4	none	synthetic target	AGATCTTACCCCTAAATATAAAGCAATTCATAGAACAATAATCTCCAGAAAAACATCGCGGAAGGGCATTAT (SEQ ID NO:20)	868300
R117H	3	none	inverter	AATCATAGCTTCTATGACCGCGGATCAAGGAGGAAT (SEQ ID NO:21)	443800
R117H	3	hex	probe/DM	CGCCGCGGAGGACTCTGCGGATTTATCT (SEQ ID NO:22)	304200
R117H	3	none	synthetic target	CTGCTAGATAAATGCGGATAGAGTGTCTCTGTTATCCGGGTGATAGGAAGCTATGATT (SEQ ID NO:23)	681700
R560T	1	none	inverter	CATGAATGACATTTACAGCAAAATCTGCTGAGCAACAATAATATGATTATCTACT (SEQ ID NO:24)	595000
R560T	1	hex	probe/ER24	ACGACGCGGAGGTTGCTAAAGAAATCTTGCT (SEQ ID NO:25)	378100
R560T	1	none	synthetic target	CAAGCAGCAAGAATTTCTTAGCAAGTGAATACGAATATGGTCTTAGCAAGCATTTGCTGTAAATGTCAATCTATCAATCTATCAATATGCTGTAAAAA (SEQ ID NO:26)	945400
3120+1G>A	2	none	inverter	GCAATTTGGATGACCTCTGCCCTCTTACCATATGACTTATCCAGT (SEQ ID NO:27)	496000
3120+1G>A	2	hex	probe/DM	CGCCGCGGAGGATGTAAAAATAGTACCCGTTAA (SEQ ID NO:28)	397500
3120+1G>A	2	none	synthetic target	AGACACTTATACGGTATCTTTTACATATCTGATGAAGTCAAAATATGGTAAAGGCGAAGGTCAATCCAAAATTTGCTATATC (SEQ ID NO:29)	984000
3659delC	2	none	inverter	GAGATGTGGCATCTTGTTGTTGTGACTTT (SEQ ID NO:30)	372300
3659delC	2	hex	probe/DM	CGCCGCGGGGTAGTACTCTTGTTGG (SEQ ID NO:31)	302800
3659delC	2	none	synthetic target	CATGCCAACAGAGGTAAACCTACAGCTCAACCAACCAACCAACAAAGATGGCGCAACTCTC (SEQ ID NO:32)	679800
A455E	1	none	inverter	CTGTAAAGATATTAATTTCAAGATAGAAAGGAGACATGTTGGT (SEQ ID NO:33)	531000
A455E	1	hex	probe/ER24	ACGACGCGGAGGAGTTGTGGATCA (SEQ ID NO:34)	298100
A455E	1	none	synthetic target	CCAGTGTCCAGCAACCTCCCAACCTGTGCTCTTCTTCTATCTTGAATTAATATCTTTCAGG (SEQ ID NO:35)	661000
1078delT	2	none	inverter	AGTGCATAGGAGGACACATAAAACACCAAT (SEQ ID NO:36)	413500
1078delT	2	hex	probe/DM	CGCCGCGAGGAGACCTGAGAGAGGAA (SEQ ID NO:37)	355400
1078delT	2	none	synthetic target	AGCCTCTCTCTCAGGTTCTGTGGTGTGTTTCTGTGCTCCCTATGCACT (SEQ ID NO:38)	533000
G551D	2	none	inverter	GCAGAGAAACACATATAGTCTTGGGAAGGTGGAAATCACACTGAGTGGAGT (SEQ ID NO:39)	628200
G551D	2	hex	probe/DM	CGCCGCGGAGTCAACGCAAGAAATTTCT (SEQ ID NO:40)	343800
G551D	2	none	synthetic target	CTTGCTAAAGAAATTTTGCTGTGCTGATCTCCACTGTGATGCCACCTTCTCCAGAACTATATGCTCTTCTGCAAACTT (SEQ ID NO:41)	883100
1148T	1	none	inverter	AAATCAACCTAAACATGATCTTCACTTGCATTCAT (SEQ ID NO:42)	432400
1148T	1	hex	probe/ER24	ACGACGCGGAGGTTGTGATGAAGGCCAAA (SEQ ID NO:43)	350200
1148T	1	none	synthetic target	CCATTTTGGCCTTCATCACACTGGAAATGCAATGAAGTATGTTAGTTGATT (SEQ ID NO:44)	643100
N1303K	2	none	inverter	CCATATTTGTGATCTCCAGCTGTTCTATAGGATGCCAT (SEQ ID NO:45)	391200
N1303K	2	hex	probe/DM	CGCCGCGGAGGCTTTTCTAAATGTTCCAGAAAAA (SEQ ID NO:46)	391200
N1303K	2	none	synthetic target	ATTATTTTCTGGAACATTTAGAAATTTGGATCCCTATGCAAGTGGAGTGCATCAAGAAATATGGAAG (SEQ ID NO:47)	87100
711+1G>T	2	none	inverter	GCCTCTGAGGATTTTCATCAATTTGTTCCAGT (SEQ ID NO:48)	
711+1G>T	2	hex	probe/DM	CGCCGCGGAGGATTCATCAATTTGTTCCAGT (SEQ ID NO:49)	
711+1G>T	2	none	synthetic target	ACCTTGAAACAAATTTGATGAATATGATGCTATGATGATTTTATCTTTTATGAGCAGTATTTGTTAAATATACAACTTGGAAAGGC (SEQ ID NO:50)	927000
711+1G>A	3	none	inverter	GCCTTTCAAATTCAGATGAGCATCAAAAGAGACTCTCAATTTTCTATTTTGGTAAAT (SEQ ID NO:51)	695000

Figure 2 cont'd

Mutation	Exemplary Pool	3' blocking group	Oligo type (Arm)	Sequence (5'-3')	ϵ_{240} M ⁻¹ cm ⁻¹
Internal control	all	none	inverted	tgctcgaacatttggagatgctcttattaccacaaatagaaaatttagagagtcactcttttagtattgctcgaatctgaaatttgaaaggcacatc (SEQ ID NO:53)	284500
Internal control	all	none	inverted	gctcgaacatttggagatgctcttattaccacaaatagaaaatttagagagtcactcttttagtattgctcgaatctgaaatttgaaaggcacatc (SEQ ID NO:54)	1010000
Internal control	all	hex	inverted	gctcgaacatttggagatgctcttattaccacaaatagaaaatttagagagtcactcttttagtattgctcgaatctgaaatttgaaaggcacatc (SEQ ID NO:55)	345000
Internal control	all	hex	inverted	gctcgaacatttggagatgctcttattaccacaaatagaaaatttagagagtcactcttttagtattgctcgaatctgaaatttgaaaggcacatc (SEQ ID NO:56)	327800
W1282X	3	none	synthetic target	gattccaatttgcgaacagcttgaaaggagctttggagtgataccacaggtgagcga (SEQ ID NO:57)	683000
3849+10kbp-T	2	none	inverted	caagagcttccatctgctgcagatataaaatggg (SEQ ID NO:58)	390000
3849+10kbp-T	2	none	inverted	cgcccgaggtgagtaagacacccctgaaa (SEQ ID NO:59)	327400
3849+10kbp-T	2	hex	synthetic target	ttcccttcagggtgctcttaccacatttataactgcgaacagatgggaagctcttg (SEQ ID NO:60)	601000
R553X	4	none	inverted	catttacgcaaatctgctgagaccacattatgattatccactctgttaagaaaattcttgctg (SEQ ID NO:61)	
R553X	4	hex	inverted	cgcccgaggtgagtaagacacccctgaaa (SEQ ID NO:62)	
R553X	4	none	synthetic target	actgagtggaagtgcaatgcaagaaattctttacgaagggtgaaataactfaattattggctgctacgaagcatttgcctggtataatg (SEQ ID NO:63)	
G542X	4	none	inverted	tccaaatttgcagagaaagacacaaatagctctcttc (SEQ ID NO:64)	
G542X	4	hex	inverted	cgcccgaggtgagtaagacacccctgaaa (SEQ ID NO:65)	
G542X	4	none	synthetic target	tgtagttccacctctcaaaagacataattgctcttctgcgaacatttggga (SEQ ID NO:66)	684000
621+1G>T	3	none	inverted	ctctcatcagatctggaatgcagatagctatgatttattatgaagacg (SEQ ID NO:67)	311900
621+1G>T	3	hex	inverted	cgcccgaggttataactctcttcgcacagg (SEQ ID NO:68)	985000
621+1G>T	3	none	synthetic target	ggcgctgtgcgaagaaatttaactctcttataataactaaactaacatagctattctcattccattccaatgtaggaagcga (SEQ ID NO:69)	467000
R334W	2	none	inverted	cggagacaaatgcagatagatgggtgggaattatctctc (SEQ ID NO:70)	336800
R334W	2	hex	inverted	cgcccgaggtgagtaagacacccctgaaa (SEQ ID NO:71)	703000
R334W	2	none	synthetic target	tgcactaatcaagggaatcatcctctgcgaaaattaccacccactctcattctgcattgttctgcg (SEQ ID NO:72)	
Internal control	all	none	inverted	tgctcgaacatttggagatgctcttattaccacaaatagaaaatttagagagtcactcttttagtattgctcgaatctgaaatttgaaaggcacatc (SEQ ID NO:73)	503500
Internal control	all	hex	inverted	tgctcgaacatttggagatgctcttattaccacaaatagaaaatttagagagtcactcttttagtattgctcgaatctgaaatttgaaaggcacatc (SEQ ID NO:74)	321200
Internal control	all	hex	inverted	tgctcgaacatttggagatgctcttattaccacaaatagaaaatttagagagtcactcttttagtattgctcgaatctgaaatttgaaaggcacatc (SEQ ID NO:75)	698200

	Hex	DMFAM
all	Hex	Y-tct-X-agg-cgg-ttt-ttc-agg-lga-gac-ctc-agg-cgg-hex (SEQ ID NO:75)
1, 2	Hex	Y-tct-X-agg-cgg-ttt-ttc-agg-lga-gac-tcc-tcc-gt-hex (SEQ ID NO:76)
5	Hex	Y-tct-X-tcc-acc-ttt-tgg-cgg-aga-gaa-gag-gac-cgg-agg-a-hex (SEQ ID NO:77)
	Hex	2NP4b/Red

X = Quencher = Z28
Y = Dye = FAM for 1055-48-08 and 1055-48-09 and Y = Z35 (or Redmond Red) for 1055-49-04

Mutation	Pool	3' blocking group	Oligo type (Arm)	Sequence (5'-3')	ϵ_{260} M ⁻¹ cm ⁻¹
delF508	delF508	none	Invader	TGATGACGCTTCTGTATCTATTCATTCATGAGAAACACA (SEQ ID NO:76)	441500
delF508	delF508	none	WT Probe	CGCGGAGGCGAAAGATGATATTTCTTAATGGT (SEQ ID NO:79)	382200
delF508	delF508	Hex	Mut Probe	AGCTGCTCGACACAATAATATTTCTTAATGGTCCA (SEQ ID NO:80)	418100
delF508	delF508	Hex	DMFAM	Y-ict-X-ggc-cgg-ttc-tcc-aga-cac-ggc-gcg-hex (SEQ ID NO:81)	347150
delF508	delF508	Hex	Wingtail/Red	Y-ict-X-tgg-gcc-ttt-tgg-cgg-aga-gat-gtc-gga-gaa-gct-hex (SEQ ID NO:82)	390400
delF508	delF508	none	Mut Target	TGCTGCGACCATTAAGAAATATCATCTCTGGTGTTCCTATGATGATACATGACAGAAGCGTCATCAAA (SEQ ID NO:83)	837500
delF508	delF508	none	Mut Target	ATGCTCGCCCATTAAGAAATATCATCTCTGGTGTTCCTATGATGATGATGACAGAAGCGTCATCAAA (SEQ ID NO:84)	828100

Mutation	Pool	3' blocking group	Oligo type (Arm)	Sequence (5'-3')
2184delA	2184delA	none	invader	CTTCCTTTTTTCCCCAAAGCTCCAGTCTGTTAAAGATTGTTTA (SEQ ID NO 85)
2184delA	2184delA	hex	MUT probe/DM	CGGCCCGAGTTTGTTCCTGCCAGGAG (SEQ ID NO 86)
2184delA	2184delA	hex	WT probe/ER24	ACGGACCGGAGTTTGTTCCTGCCAGGAG (SEQ ID NO 87)

X = Quencher = Z28

Y = Dye = FAM for 1055-48-08 and Y = Z35 (or Redmond Red) for 1144-16-02

711+1G>	2	hex	probe/ER24	ACGACGCGGAGAAATCATCAATTTGTTACAGG (SEQ ID NO:88)
3849+10kb	2	hex	probe/ER24	ACGACGCGGAGTGAGTAAGACACCGCTGAAA (SEQ ID NO:89)